



TGM3 gene

transglutaminase 3

Normal Function

The *TGM3* gene provides instructions for making an enzyme called transglutaminase 3. This enzyme is found in certain skin cells called keratinocytes and corneocytes, as well as in various structures that make up scalp hair, including the root and strand (shaft).

Transglutaminase 3 helps proteins attach (bind) to each other at specific protein building blocks (amino acids). Specifically, transglutaminase 3 helps bind proteins together at their glutamine and lysine amino acids. This binding forms stabilizing cross-links between proteins. These protein cross-links provide strength and structure to cells, particularly skin and hair cells.

Health Conditions Related to Genetic Changes

uncombable hair syndrome

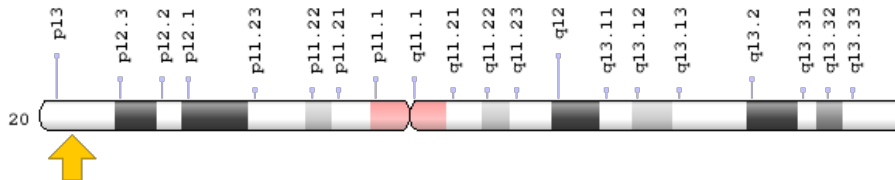
At least one mutation in the *TGM3* gene has been found to cause uncombable hair syndrome. This condition is characterized by dry, frizzy, blond scalp hair that cannot be combed flat. This condition usually improves over time, and by adolescence individuals with uncombable hair syndrome have hair that lies flat and has normal or nearly normal texture.

The *TGM3* gene mutation that has been identified leads to a premature stop signal in the instructions used to make transglutaminase 3, resulting in an abnormally short enzyme with severely reduced activity. A shortage (deficiency) of functional enzyme impairs cross-linking between certain proteins. Particularly, the hair shaft protein trichohyalin cannot bind to other trichohyalin proteins or to molecules called keratin intermediate filaments. These proteins and molecules need to bind to each other to form the cross-links that give the hair shaft its cylindrical shape. Because transglutaminase 3 cannot facilitate these cross-links, the cross-section of the hair shaft becomes triangular, heart-shaped, or flat. These angular hair shafts result in frizzy hair that will not lie flat, which is typical of uncombable hair syndrome.

Chromosomal Location

Cytogenetic Location: 20p13, which is the short (p) arm of chromosome 20 at position 13

Molecular Location: base pairs 2,295,967 to 2,341,079 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- E polypeptide, protein-glutamine-gamma-glutamyltransferase
- protein-glutamine gamma-glutamyltransferase E
- TG(E)
- TGase-3
- TGase E
- TGE
- transglutaminase E

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Cutaneous Appendages
<https://www.ncbi.nlm.nih.gov/books/NBK10037/#A2933>
- Madame Curie Bioscience: Molecular Mechanisms of Embryonic and Adult Hair Development
<https://www.ncbi.nlm.nih.gov/books/NBK6571/#A62117>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGM3%5BTIAB%5D%29+OR+%28transglutaminase+3%5BTIAB%5D%29%29+OR+%28TGase-3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- TRANSGLUTAMINASE 3
<http://omim.org/entry/600238>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TGM3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGM3%5Bgene%5D>
- HGNC Gene Family: Transglutaminases
<http://www.genenames.org/cgi-bin/genefamilies/set/773>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11779
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7053>
- UniProt
<http://www.uniprot.org/uniprot/Q08188>

Sources for This Summary

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